

Claims

- [c1] 1. A method for providing information about biological molecules, comprising the acts of:
- receiving a user selection of one or more probe set identifiers that identify probe sets of one or more synthesized or spotted probe arrays capable of detecting the biological molecules;
- determining one or more alternative splice variants based at least in part upon the one or more probe set identifiers, wherein at least one of the probe arrays or at least one of the probe sets is constructed and arranged to detect or measure any one or any combination of gene expression, genotype, SNP, haplotype, or targets including antibodies, cell membrane receptors, monoclonal antibodies and antisera reactive with specific antigenic determinants, drugs, oligonucleotides, nucleic acids, peptides, proteins, cofactors, lectins, sugars, polysaccharides, cells, cellular membranes, or organelles;
- correlating at least one alternative splice variant with at least one annotation datum; and
- providing to the user, over a network, a graphical representation of the at least one alternative splice variant and the correlated annotation datum.
- [c2] 2. The method of claim 1, wherein:
- one or more of the probe arrays is constructed and arranged to diagnose a disease or medical condition.
- [c3] 3. A method, comprising the acts of:
- determining one or more alternative splice variants based at least in part upon one or more probe set identifiers that identify probe sets capable of detecting biological molecules;
- correlating at least one alternative splice variant with at least one annotation datum; and
- enabling for display a representation of the at least one alternative splice variant and the correlated annotation datum.
- [c4] 4. The method of claim 3, further comprising the act of:

receiving from a user a selection of the one or more probe set identifiers.

- [c5] 5. The method of claim 4 , wherein:
the act of receiving includes the acts of the user originating the selection from a user computer, transmission of the selection over the Internet, and receipt of the selection at an Internet server.

- [c6] 6. The method of claim 3 , wherein:
the probe sets include probes of a synthesized or spotted probe array.

- [c7] 7. The method of claim 3 , wherein:
the probe sets include probes disposed on or in a support comprising beads, resins, gels, or microspheres.

- [c8] 8. The method of claim 3 , wherein:
the probe sets include probes of a probe array, wherein the probe sets or probe array are constructed and arranged to detect or measure any one or any combination of gene expression, genotype, SNP, haplotype, or targets including antibodies, cell membrane receptors, monoclonal antibodies and antisera reactive with specific antigenic determinants, drugs, oligonucleotides, nucleic acids, peptides, proteins, cofactors, lectins, sugars, polysaccharides, cells, cellular membranes, or organelles.

- [c9] 9. The method of claim 3 , wherein:
the act of determining includes model fitting.

- [c10] 10. The method of claim 3 , wherein:
the act of determining includes verifying using protein domain data.

- [c11] 11. The method of claim 3 , wherein:
the act of correlating includes correlating the at least one alternative splice variant with a gene and correlating the gene with the at least one annotation datum.

- [c12] 12. The method of claim 3 , wherein:
the act of correlating includes correlating the at least one alternative splice variant with at least one other alternative splice variant of their common gene

and correlating the at least one other alternative splice variant with the at least one annotation datum.

- [c13] 13. The method of claim 3, wherein:
the representation of the at least one alternative splice variant or of the at least one annotation datum is constructed and arranged to enable semantic zooming wherein magnification is determined, at least in part, on a user zoom selection.
- [c14] 14. The method of claim 13, wherein:
the annotation datum includes sequence information displayed on a sequence axis, and the semantic zooming is along a single dimension corresponding to the sequence axis.
- [c15] 15. The method of claim 13, wherein:
the representation of the at least one alternative splice variant or of the at least one annotation datum is organized into a plurality of adjustable tiers that are constructed and arranged for display so as to be capable of being collapsed, moved, or hidden in response to user tier selection.
- [c16] 16. The method of claim 3, wherein:
the representation of the at least one annotation datum is constructed and arranged for display based, at least in part, on a user selection of one or more of a genomic, primary-transcript, mRNA, or protein display type.
- [c17] 17. The method of claim 3, wherein:
the at least one annotation datum includes any one or any combination of genomic sequence; presence or relative abundance of alternative splice variants; exon arrangement, content, or sequence; intron arrangement, content, or sequence; frequency of exon usage in two or more of the alternative splice variants; isoform identification; primary transcript, mRNA or other RNA identification, function, structure, or sequence; protein, protein domain, or protein motif identification, function, structure, or sequence; gene identification, function, structure, or sequence for a gene corresponding to the at least one alternative splice variant; one or more start or stop sites; 5' and 3' untranslated regions; coding regions; protein-based annotations of the coding

regions; start and stop codons; 5' transcriptional control elements; 3' polyadenylation signals; splice site boundaries; probe arrangement, content, or sequence; or expression level data corresponding to one or more probes of the probe sets.

- [c18] 18. The method of claim 3 , wherein:
the act of enabling for display includes aligning the representation of a first alternative splice variant with a second alternative splice variant, wherein the first and second alternative splice variants are variants of a same gene.
- [c19] 19. The method of claim 18 , wherein:
the aligning is based, at least in part, on sequences of the first and second alternative splice variants compared to genomic sequence or sequence of the same gene.
- [c20] 20. The method of claim 3 , wherein:
the act of enabling for display includes graphically associating the alternative splice variant and the annotation datum.
- [c21] 21. The method of claim 3 , wherein:
the act of enabling for display includes enabling separate display of a plurality of annotation data in a plurality of panes of a single graphical user interface.
- [c22] 22. The method of claim 3 , further comprising the acts of:
receiving a selection by a user of the one or more probe set identifiers; and
providing to the user the representation of the at least one alternative splice variant and the correlated annotation datum.
- [c23] 23. The method of claim 22 , wherein:
the acts of receiving and providing are accomplished, at least in part, over a network including any one or any combination of the Internet, an intranet, or a local area network.
- [c24] 24. The method of claim 3 , wherein:
the probe sets comprise probes constructed and arranged to detect mRNA expression.

- [c25] 25. The method of claim 3 , wherein:
the probes comprise exon probes or junction probes.
- [c26] 26. The method of claim 3 , further comprising the act of:
receiving one or more hybridization intensity values corresponding to the one
or more probe set identifiers, wherein the hybridization intensity values are
produced from biological probe array experiments.
- [c27] 27. The method of claim 26 , wherein:
the act of determining is based, at least in part, on the one or more probe set
identifiers and their corresponding hybridization intensity values.
- [c28] 28. A system comprising:
an alternative splice variant evaluator constructed and arranged to determine
one or more alternative splice variants based at least in part upon one or more
probe set identifiers that identify probe sets capable of detecting biological
molecules;
an alternative splice variant data storage and annotation data correlator
constructed and arranged to correlate at least one alternative splice variant with
at least one annotation datum; and
a user-service manager constructed and arranged to enable for display a
representation of the at least one alternative splice variant and the correlated
annotation datum.
- [c29] 29. The system of claim 28 , further comprising:
an input manager constructed and arranged to receive from a user a selection of
the one or more probe set identifiers.
- [c30] 30. The system of claim 29 , wherein:
the input manager receives the user selection over the Internet.
- [c31] 31. The system of claim 28 , wherein:
the probe sets include probes of a probe array constructed and arranged to
detect or measure any one or any combination of gene expression, genotype,
SNP, haplotype, or targets including antibodies, cell membrane receptors,
monoclonal antibodies and antisera reactive with specific antigenic

determinants, drugs, oligonucleotides, nucleic acids, peptides, proteins, cofactors, lectins, sugars, polysaccharides, cells, cellular membranes, or organelles.

- [c32] 32. The system of claim 28 , wherein:
the representation of the at least one alternative splice variant or of the at least one annotation datum is constructed and arranged to enable semantic zooming wherein magnification is determined, at least in part, on a user zoom selection.
- [c33] 33. The system of claim 28 , wherein:
the representation of the at least one annotation datum is constructed and arranged for display based, at least in part, on a user selection of one or more of a genomic, primary-transcript, mRNA, or protein display type.
- [c34] 34. The system of claim 28 , wherein:
the at least one annotation datum includes any one or any combination of genomic sequence; presence or relative abundance of alternative splice variants; exon arrangement, content, or sequence; intron arrangement, content, or sequence; frequency of exon usage in two or more of the alternative splice variants; isoform identification; primary transcript, mRNA or other RNA identification, function, structure, or sequence; protein, protein domain, or protein motif identification, function, structure, or sequence; gene identification, function, structure, or sequence for a gene corresponding to the at least one alternative splice variant; one or more start or stop sites; 5' and 3' untranslated regions; coding regions; protein-based annotations of the coding regions; start and stop codons; 5' transcriptional control elements; 3' polyadenylation signals; splice site boundaries; probe arrangement, content, or sequence; or expression level data corresponding to one or more probes of the probe sets.
- [c35] 35. The system of claim 28 , wherein:
the user-service manager further is constructed and arranged to align the representation of a first alternative splice variant with a second alternative splice variant, wherein the first and second alternative splice variants are variants of a same gene.

- [c36] 36. The system of claim 35 , wherein:
the aligning is based, at least in part, on sequences of the first and second alternative splice variants compared to genomic sequence or sequence of the same gene.
- [c37] 37. The system of claim 28 , wherein:
the user-service manager further is constructed and arranged to graphically associate the alternative splice variant and the annotation datum.
- [c38] 38. The system of claim 28 , further comprising:
an input manager constructed and arranged to receive from a user over a first network a selection of the one or more probe set identifiers; and
an output manager constructed and arranged to send to the user over a second network the representation of the at least one alternative splice variant and the correlated annotation datum.
- [c39] 39. The system of claim 38 , wherein:
the first and second networks are a same network or different networks including any one or any combination of the Internet, an intranet, or a local area network.
- [c40] 40. The system of claim 28 , wherein:
the probe sets comprise probes constructed and arranged to detect mRNA expression.
- [c41] 41. The system of claim 28 , wherein:
the probes comprise exon probes or junction probes.
- [c42] 42. The system of claim 29 , wherein:
the input manager further is constructed and arranged to receive one or more hybridization intensity values corresponding to the one or more probe set identifiers, wherein the hybridization intensity values are produced from biological probe array experiments; and
the alternative splice variant evaluator further is constructed and arranged to determine the one or more alternative splice variants based, at least in part, on the one or more probe set identifiers and their corresponding hybridization

intensity values.

[c43] 43. A genomic web portal, comprising:
an input manager constructed and arranged to receive from a user over the Internet a selection of one or more probe set identifiers that identify probe sets capable of detecting biological molecules and one or more hybridization intensity values corresponding to the one or more probe set identifiers, wherein the hybridization intensity values are produced from biological probe array experiments;
an alternative splice variant evaluator constructed and arranged to determine one or more alternative splice variants based at least in part upon one or more probe set identifiers and their corresponding hybridization intensity values;
an alternative splice variant data storage and annotation data correlator constructed and arranged to correlate at least one alternative splice variant with at least one annotation datum;
a user-service manager constructed and arranged to enable for display a representation of the at least one alternative splice variant and the correlated annotation datum; and
an output manager constructed and arranged to send to the user over the Internet the representation of the at least one alternative splice variant and the correlated annotation datum.

[c44] 44. The genomic web portal of claim 43 , wherein:
the representation of the at least one annotation datum is constructed and arranged for display based, at least in part, on a user selection of one or more of a genomic, primary-transcript, mRNA, or protein display type.

[c45] 45. The genomic web portal of claim 43 , wherein:
the at least one annotation datum includes any one or any combination of genomic sequence; presence or relative abundance of alternative splice variants; exon arrangement, content, or sequence; intron arrangement, content, or sequence; frequency of exon usage in two or more of the alternative splice variants; isoform identification; primary transcript, mRNA or other RNA identification, function, structure, or sequence; protein, protein domain, or

protein motif identification, function, structure, or sequence; gene identification, function, structure, or sequence for a gene corresponding to the at least one alternative splice variant; one or more start or stop sites; 5' and 3' untranslated regions; coding regions; protein-based annotations of the coding regions; start and stop codons; 5' transcriptional control elements; 3' polyadenylation signals; splice site boundaries; probe arrangement, content, or sequence; or expression level data corresponding to one or more probes of the probe sets.